

# Giancarlo Deidda

## List of Publications by Year in descending order

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23  
papers

1,237  
citations

567281

15  
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642732

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docs citations

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#	ARTICLE	IF	CITATIONS
1	Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. <i>European Journal of Neurology</i> , 2020, 27, 2604-2615.	3.3	16
2	Digenic Inheritance of Shortened Repeat Units of the D4Z4 Region and a Loss-of-Function Variant in SMCHD1 in a Family With FSHD. <i>Frontiers in Neurology</i> , 2018, 9, 1027.	2.4	8
3	Estrogens enhance myoblast differentiation in facioscapulohumeral muscular dystrophy by antagonizing DUX4 activity. <i>Journal of Clinical Investigation</i> , 2017, 127, 1531-1545.	8.2	46
4	Allele-specific DNA hypomethylation characterises FSHD1 and FSHD2. <i>Journal of Medical Genetics</i> , 2016, 53, 348-355.	3.2	54
5	Highly efficient, in vivo optimized, archaeal endonuclease for controlled RNA splicing in mammalian cells. <i>FASEB Journal</i> , 2013, 27, 3466-3477.	0.5	2
6	ARCHAEA-EXPREs targeting of $\beta$ -tubulin 4 mRNA: a model for high-specificity trans-splicing. <i>FASEB Journal</i> , 2010, 24, 2976-2984.	0.5	3
7	FRG2, an FSHD candidate gene, is transcriptionally upregulated in differentiating primary myoblast cultures of FSHD patients. <i>Journal of Medical Genetics</i> , 2004, 41, 826-836.	3.2	76
8	An archaeal endoribonuclease catalyzes cis- and trans- nonspliceosomal splicing in mouse cells. <i>Nature Biotechnology</i> , 2003, 21, 1499-1504.	17.5	24
9	Interchromosomal repeat array interactions between chromosomes 4 and 10: a model for subtelomeric plasticity. <i>Human Molecular Genetics</i> , 2000, 9, 2879-2884.	2.9	95
10	De Novo Facioscapulohumeral Muscular Dystrophy: Frequent Somatic Mosaicism, Sex-Dependent Phenotype, and the Role of Mitotic Transchromosomal Repeat Interaction between Chromosomes 4 and 10. <i>American Journal of Human Genetics</i> , 2000, 66, 26-35.	6.2	136
11	Progress in the molecular diagnosis of facioscapulohumeral muscular dystrophy and correlation between the number of KpnI repeats at the 4q35 locus and clinical phenotype. <i>Annals of Neurology</i> , 1999, 45, 751-757.	5.3	263
12	Molecular analysis of 4q35 rearrangements in facioscapulohumeral muscular dystrophy (FSHD): application to family studies for a correct genetic advice and a reliable prenatal diagnosis of the disease. <i>Neuromuscular Disorders</i> , 1999, 9, 190-198.	0.6	26
13	A new dosage test for subtelomeric 4;10 translocations improves conventional diagnosis of facioscapulohumeral muscular dystrophy (FSHD). <i>Journal of Medical Genetics</i> , 1999, 36, 823-8.	3.2	22
14	Sequence Homology between 4qter and 10qter Loci Facilitates the Instability of Subtelomeric KpnI Repeat Units Implicated in Facioscapulohumeral Muscular Dystrophy. <i>American Journal of Human Genetics</i> , 1998, 63, 181-190.	6.2	60
15	Inter- and intrachromosomal sub-telomeric rearrangements on 4q35: implications for facioscapulohumeral muscular dystrophy (FSHD) aetiology and diagnosis. <i>Human Molecular Genetics</i> , 1998, 7, 1207-1214.	2.9	96
16	Direct detection of 4q35 rearrangements implicated in facioscapulohumeral muscular dystrophy (FSHD). <i>Journal of Medical Genetics</i> , 1996, 33, 361-365.	3.2	129
17	Analysis of $\beta$ -thalassaemia mutations in the United Arab Emirates provides evidence for recurrent origin of the IVSINT 5 (G-C) mutation. <i>Human Mutation</i> , 1995, 5, 327-328.	2.5	9
18	Physical Mapping Evidence for a Duplicated Region on Chromosome 10qter Showing High Homology with the Facioscapulohumeral Muscular Dystrophy Locus on Chromosome 4qter. <i>European Journal of Human Genetics</i> , 1995, 3, 155-167.	2.8	89

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19	Chromosome 4q35 haplotypes and DNA rearrangements segregating in affected subjects of 19 Italian families with facioscapulohumeral muscular dystrophy (FSHD). <i>Human Genetics</i> , 1994, 94, 367-374.	3.8	12
20	A new $\hat{\beta}^2$ -thalassaemia frameshift mutation detected by PCR after selective hybridization to immobilized oligonucleotides. <i>British Journal of Haematology</i> , 1991, 79, 90-92.	2.5	7
21	Molecular characterization of $\hat{\beta}^2$ -thalassemia mutations in Egypt. <i>Human Genetics</i> , 1990, 85, 272-274.	3.8	39
22	A New $\hat{\beta}^2$ -Thalassemia Mutation Produced by a Single Nucleotide Substitution in the Conserved Dinucleotide Sequence of the IVS-I Consensus Acceptor Site (A $\hat{g}\hat{a}\hat{t}$ 'AA). <i>Hemoglobin</i> , 1990, 14, 431-440.	0.8	16
23	Frequency and molecular types of deletional $\hat{\beta}^{\pm}$ -thalassemia in Egypt. <i>Human Genetics</i> , 1989, 81, 211-213.	3.8	9